

Application No. 10/672,937
Amendment dated June 27, 2006
Response to Office Action dated December 27, 2006

Amendments to the Specification:

Please replace the Title of the application with the following title:

SYSTEM AND METHOD FOR MITOCHONDRIAL DNA AUTOSCORING SYSTEM OF GENETIC SEQUENCES.

Please replace paragraph [0008] with the following amended paragraph:

[0008] In still other embodiments, the system may be adapted for use with automated sequencers or base calling systems, sequence assembly systems, and post-processing systems. Examples of base calling systems and methods known in the art, include, but are not limited to, phred (Ewing, B., et al. Genome Research 8:175-185, 1998). Examples of sequence assembly systems and methods known in the art, include but are not limited to phrap (www.phrap.org). In certain embodiments, the post-processing systems and modules automate data analysis by implementing rules typically embodied in procedures for manual data review. Examples of rules typically embodied in standard procedures for manual review include, but are not limited to, coverage requirements, definitions of background versus mixed base calls, detection of mixtures, and heteroplasmic site rules.

Please replace paragraph [0023] with the following amended paragraph:

[0023] Figure 5 entitled Table 1 describes the function performed by various software programs that may be integrated into the autoanalysis approach.

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Please replace paragraph [0024] with the following amended paragraph:

[0024] ~~Figure 6~~ ~~Table 2~~ illustrates various validation statistics comparing the use of the autoanalysis methods with conventional manual-based analysis methods.

Please replace paragraph [0099] with the following amended paragraph:

[0099] As previously indicated, in certain embodiments, the automated data analysis system may include software programs and algorithms developed by Applera Corporation. For example, some embodiments of the system include the following programs: BlastParse.pl; find_bad_traces_from_blast_report.pl; mark_substitution_heteroplasmic.pl; determineReadTypes.pl; extract_SE_consensus.pl; seq2delta_vs; compute_coverage.pl; calculate_coverage_mitotype.pl; count_hv1_deletes.pl; count_hv1_inserts.pl; border_index.pl; generate_hv_mask_fasta_files.pl; fix_mitotype_reporting_range.pl; flip_fasta.pl. A brief description of each of these programs is provided in Table 1 the table shown in Figure 5.

Please replace paragraph [0111] with the following amended paragraph:

[0111] ~~As shown in Table 2~~ the table shown in Fig. 6, the false negative results indicated that the system provided 99.88% specificity and 99.99% negative predictive value on the test sample sets. The sensitivity evaluation indicated that the ability of the system to accurately identify ambiguous positions is reasonable. When comparing the number of false positives versus the number of false negatives, and the positive predictive value, this analysis demonstrates that it would be more likely for the automated data analysis system to call an unambiguous base as

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ambiguous than vice versa.

Please replace paragraph [0115] with the following amended paragraph:

[0115] The results, summarized in Table 2 the table shown in Fig. 6, demonstrate that the automated data analysis system provided 99.27% specificity and 99.99% negative predictive value on the test sample sets. The sensitivity evaluation indicated that the ability of the automated system to accurately identify ambiguous positions is reasonable. When comparing the number of false positives versus the number of false negatives, and the positive predictive value, this analysis demonstrates that it would be more likely for the automated data analysis system to call an unambiguous base as ambiguous than vice versa.